

The most definitive treatment to date is liver transplantation; this replaces a liver that lacks LDL receptors with one having the normal complement. There is only one reported case in the literature: a child with familial hypercholesterolaemia and severe coronary artery disease, who had a combined heart and liver transplant⁷. She did very well, with a marked reduction in serum cholesterol and enhanced LDL clearance from the plasma postoperatively.

Our patient is being considered for plasmapheresis or possible liver transplantation in the future.

References

- 1 Frederickson DS. Plasma lipid abnormalities and cutaneous and subcutaneous xanthomas. In: Fitzpatrick TB, ed. *Dermatology in General Medicine*. New York: McGraw-Hill, 1979:1112-25
- 2 Brown MS, Goldstein JL. Receptor mediated control of cholesterol metabolism. *Science* 1976;191:150-3

- 3 West R, Gibson P, Lloyd J. Treatment of homozygous familial hypercholesterolaemia: an informative sibship. *Br Med J* 1985;291:1079-80
- 4 Thompson GR, Lowenthal R, Myant NB. Plasma exchange in the management of homozygous familial hypercholesterolaemia. *Lancet* 1975;i:1208-11
- 5 Thompson GR, Miller JP, Breslow JL. Improved survival of patients with homozygous familial hypercholesterolaemia treated with plasma exchange. *Br Med J* 1985;291:1671-3
- 6 Starzl TH, Chase HP, Ahrens EH, et al. Portacaval shunt in patient with familial hypercholesterolaemia. *Ann Surg* 1983;198:273-81
- 7 Bilheimer DW, Goldstein JL, Grundy SM, Starzl TE, Brown MS. Liver transplantation to provide low density lipoprotein receptors and lower plasma cholesterol in a child with homozygous familial hypercholesterolaemia. *N Engl J Med* 1984;311:1658-63

(Accepted 14 July 1986. Dr Phillips is now at the Boston University Medical Center, Boston, Massachusetts, USA)

Perianal pain and swelling due to a pre-coccygeal tumour

T Ward MB S J Snooks MD FRCS
R J Croft MChir FRCS *North Middlesex Hospital,*
London N18

Keywords: glioma, extramedullary; perianal pain

The differential diagnosis of coccygeal swellings includes congenital abnormalities, subcutaneous tumours such as lipomas, fractures or tumours of the coccyx itself; perianal lesions such as abscesses and fistulae and pilonidal sinuses. All of these lesions may present with perianal and/or coccygeal discomfort. Quite often perianal pain has no obvious cause and management is therefore difficult¹. A case is now reported where a coccygeal tumour was the cause of perianal pain and coccygeal swelling.

Case report

A 34-year-old Caucasian housewife, who was otherwise fit and well, presented with a two-year history of a lump in the coccygeal region which she had initially noticed when bending down in a telephone booth to retrieve a dropped coin. This lump had increased in size and was uncomfortable on sitting down, so much so that the patient complained that she felt as though she was sitting on a melon when she had a bath. She was not incontinent of urine or faeces and was not constipated.

On examination there was a 4 × 3 cm round, non-fluctuant swelling in the coccygeal region which was non-tender and did not transilluminate. No fissure, fistulae or sinuses were demonstrated and digital rectal examination was normal. Neurological examination of the upper and lower limbs revealed normal reflexes, sensation and motor function. No neurological abnormality was found of the pelvic floor muscu-

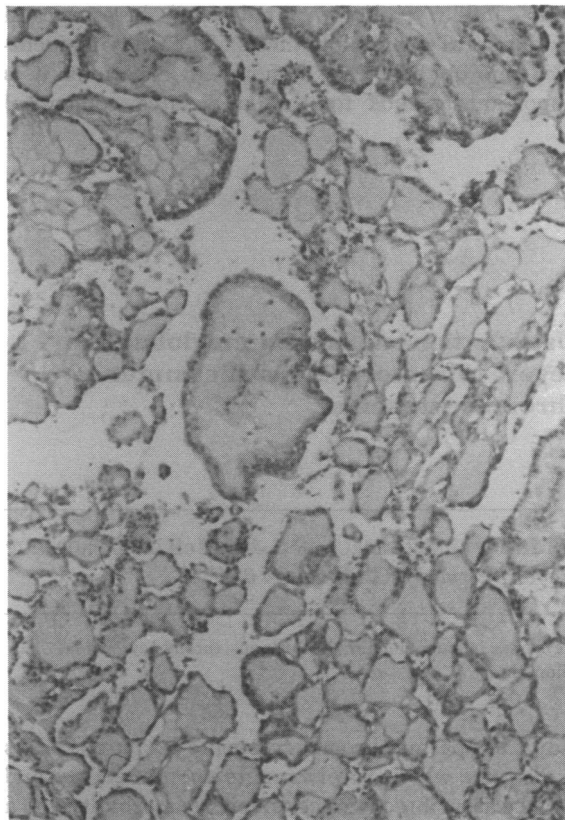


Figure 1. Extramedullary glioma

lature. Radiological examination of the lumbosacral spine and coccyx was normal.

At operation a 4 cm soft encapsulated mass was found which was unattached to the coccyx. Histology of this tumour showed an encapsulated lesion composed of papillary structures covered by cuboidal epithelium with areas of myxoid stroma. No mitotic figures were seen, and the features were consistent with a myxopapillary ependymoma – a type of extramedullary glioma (Figure 1). Postoperatively the patient did not have any abnormal neurological signs, and lumbosacral myelography was normal.

Case presented
to Clinical
Section,
10 October 1986

0141/0768/87/
010651-02/\$02.00/0
© 1987
The Royal
Society of
Medicine

Discussion

Gliomas are tumours of nervous tissue origin, composed of cells closely resembling astrocytes, oligodendrocytes or ependymal cells, the respective tumours being known as astrocytomas, oligodendrogliomas and ependymomas. Anaplastic forms also occur which contain pleomorphic, less well differentiated cell types.

Extramedullary (or heterotopic) gliomas occur outside the brain or spinal cord. They are most commonly ependymomas. Cooper *et al.*² have previously reported a series of 15 extramedullary gliomas which occurred along the spinal axis, but without any apparent attachment thereto, and which had not arisen by seeding from a primary intramedullary glial or intracerebral neoplasm. Characteristic features of extramedullary gliomas are: (1) lack of attachment to the central nervous system and absence of signs of a primary neoplastic process within the brain or spinal cord; (2) the encapsulated appearance of the tumour; (3) location of the tumour along the neuraxis and predominantly in the lumbosacral region². The patient described in this report exhibits all of these features.

Often extramedullary gliomas are associated with congenital abnormalities such as myelomeningocele, spina bifida, club foot, undescended testes and lumbosacral spondylolisthesis². Thus, having identified one of these rare tumours in a patient, it is important to look for any associated congenital

abnormalities as well as performing a careful neurological assessment.

The mechanism of formation of extramedullary gliomas is considered to be a 'pinching off' of glial tissue from the neural tube, in the same way that dermoid and epidermoid tumours arise from aberrant cutaneous tissue which is caught in the closure of the neural tube, thus enabling cutaneous tissue to come to rest in the intramedullary portion of the neuraxis. The majority of such heterotopic glial and dermoid tumours occur in this lumbosacral region, and Humphrey³ concluded that this was because it is at the caudal end of the embryo, where disparities in the developmental rates of related structures most often lead to abnormalities.

Tumours of neural origin should be considered when no obvious cause of perianal discomfort or pain is found in the sacrococcygeal region.

References

- 1 Neill M, Swash M. Chronic perianal pain – an unsolved problem. *J R Soc Med* 1982;75:96–101
- 2 Cooper IS, Kenohan JW, Craig WMcK. Tumours of the spinal cord – primary extramedullary gliomas. *Surg Gynecol Obstet* 1951;92:183
- 3 Humphrey T. Sensory ganglion cells within the central canal of the embryonic human spinal cord. *J Comp Neurol* 1947;86:1–21

(Accepted 21 July 1986)

Use of intravenous immunoglobulin in severe refractory idiopathic immune thrombocytopenia

D Maharaj MRCP MRCPATH **J H McKillop** PhD MRCP
Royal Infirmary, Glasgow G4 0SF

Keywords: immunoglobulin, intravenous; thrombocytopenia, idiopathic

Adult idiopathic thrombocytopenic purpura (ITP) is a disorder which rarely remits spontaneously and may be associated with considerable morbidity and a mortality rate of 5%¹. The management of refractory cases with sufficiently severe thrombocytopenia to cause bleeding often poses a major clinical problem, as transfused platelets rarely reduce the bleeding tendency or result in a rise in the circulating platelet count. We report the successful use of high-dose intravenous immunoglobulin in increasing the platelet count in three cases of severe idiopathic thrombocytopenic purpura, in whom treatment with corticosteroids was unsuccessful or contraindicated.

Case reports

Case 1: A 65-year-old woman presented to her general practitioner with a three-week history of bruising of the lower limbs. For five years she had been treated

with Aldactide 50 (hydroflumethiazide 50 mg and spironolactone 50 mg) two tablets daily for mild hypertension. She had a perforated duodenal ulcer one year previously and was on a maintenance dose of cimetidine 400 mg at night. Physical examination revealed scattered petechiae on the lower limbs and an aortic aneurysm was palpable in the epigastrium. The platelet count was reduced at $30 \times 10^9/l$, and this was thought to be drug-induced thrombocytopenia. All treatment was withdrawn but three months later her platelet count was unchanged and she was referred for further investigation of thrombocytopenia and abdominal aneurysm.

Haematological investigation showed a haemoglobin concentration of 12.5 g/dl, white cell count of $5.4 \times 10^9/l$ and platelet count $34 \times 10^9/l$. Serum B₁₂ and folate and red cell folate were normal. Rheumatoid factor, antinuclear factor and viral screen were negative. Platelet-associated IgG (PAIgG) and IgM (PAIgM) levels were elevated. Sternal bone marrow aspirate showed an increased number of megakaryocytes. Abdominal ultrasound revealed a greatly enlarged aortic aneurysm extending from the coeliac axis to the iliac vessels.

Urgent surgical repair of the aneurysm was required and a normal platelet count was considered necessary prior to operation. High-dose steroids were contraindicated in view of her history of peptic ulceration. High-dose immunoglobulin (1.0 g/kg/day; total dose 62 g/day) was infused intravenously over two days. Within 24 hours of infusion the platelet count rose to a normal level. Splenectomy and repair